



ACVR1 gene

activin A receptor type 1

Normal Function

The *ACVR1* gene provides instructions for making the activin receptor type I protein, which is a member of a protein family called bone morphogenetic protein (BMP) type I receptors. BMP receptors span the cell membrane, so that one end of the protein remains inside the cell and the other end projects from the outer surface of the cell. This arrangement allows receptors to receive signals from outside the cell and transmit them inside to affect cell development and function.

Activin receptor type I is found in many tissues of the body including skeletal muscle and cartilage. It helps to control the growth and development of the bones and muscles, including the gradual replacement of cartilage by bone (ossification). This process occurs in normal skeletal maturation from birth to young adulthood.

Activin receptor type I is normally activated at appropriate times by molecules called ligands. Activation may occur when these ligands, such as BMPs, attach (bind) to the receptor or to other proteins with which it forms a complex. A protein called FKBP12 can inhibit activin receptor type I by binding to the receptor and preventing inappropriate (leaky) activation in the absence of ligand.

Health Conditions Related to Genetic Changes

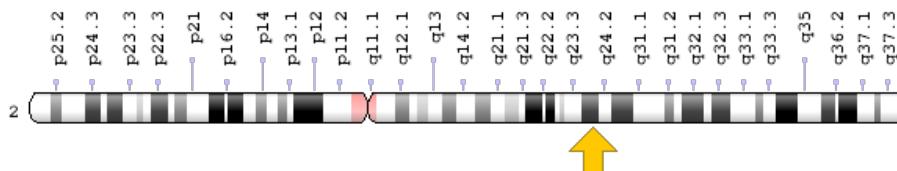
fibrodysplasia ossificans progressiva

All individuals with a definite diagnosis of fibrodysplasia ossificans progressiva have a mutation in which the protein building block (amino acid) histidine is substituted for the amino acid arginine at position 206 of the *ACVR1* protein (written as Arg206His or R206H). Researchers believe that under certain conditions this mutation may change the shape of the receptor. This shape change may disrupt the binding of an inhibitor protein such as FKBP12 or interfere with other mechanisms that control activation. As a result, the receptor may be constantly activated (constitutive activation), even in the absence of ligands. Constitutive activation of the receptor causes overgrowth of bone and cartilage and fusion of joints, resulting in the signs and symptoms of fibrodysplasia ossificans progressiva.

Chromosomal Location

Cytogenetic Location: 2q24.1, which is the long (q) arm of chromosome 2 at position 24.1

Molecular Location: base pairs 157,736,446 to 157,875,880 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- activin A receptor type I
- activin A receptor, type I
- activin A receptor, type II-like kinase 2
- activin A type I receptor
- activin A type I receptor precursor
- ActR-IA protein, human
- ACTRI
- ACVR1_HUMAN
- ACVR1A
- ACVRLK2
- ALK2
- hydroxyalkyl-protein kinase
- SKR1

Additional Information & Resources

Genetic Testing Registry

- GTR: Genetic tests for ACVR1
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=90%5Bgeneid%5D>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ACVR1%5BTIAB%5D%29+OR+%28ALK2%5BTIAB%5D%29+OR+%28SKR1%5BTIAB%5D%29+OR+%28ACTRI%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- ACTIVIN A RECEPTOR, TYPE I
<http://omim.org/entry/102576>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/ACVR1ID564ch2q24.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ACVR1%5Bgene%5D>
- HGNC Gene Family: Type 1 receptor serine/threonine kinases
<http://www.genenames.org/cgi-bin/genefamilies/set/345>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=171
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/90>
- UniProt
<http://www.uniprot.org/uniprot/Q04771>

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